

Claims

1. A nucleic acid molecule encoding a (poly)peptide co-segregating in mutated form with Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy (APECED) which is
- (a) a nucleic acid molecule comprising a nucleic acid molecule encoding the (poly)peptide having the amino acid sequence of Fig. 2A;
 - (b) a nucleic acid molecule comprising the nucleic acid molecule having the nucleotide sequence of Fig. 2A that encodes the amino acid sequence of Fig. 2A;
 - (c) a nucleic acid molecule hybridizing to the nucleic acid molecule of (a) or (b); or
 - (d) a nucleic acid molecule which is degenerate to the nucleic acid molecule of (c).
2. The nucleic acid molecule of claim 1, wherein said polypeptide has the function of a transcription factor or a transcription-associated factor.
3. The nucleic acid molecule of claim 1 or 2, wherein said polypeptide comprises two double-paired zinc finger motifs.
4. A nucleic acid molecule which is a mammalian homologue of the nucleic acid molecule of any one of claims 1 to 3.
5. The nucleic acid molecule of claim 4 which is a murine homologue.

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6. The nucleic acid molecule of claim 5 which is
- (a) a nucleic acid molecule comprising a nucleic acid molecule encoding the (poly)peptide having the amino acid sequence of Fig. 14;
 - (b) a nucleic acid molecule comprising the nucleic acid molecule having the nucleotide sequence of Fig. 14 that encodes the amino acid sequence of Fig. 14;
 - (c) a nucleic acid molecule hybridizing to the nucleic acid molecule of (a) or (b); or
 - (d) a nucleic acid molecule which is degenerate to the nucleic acid molecule of (c).
7. A nucleic acid molecule deviating by at least one mutation from the nucleic acid molecule of claim 1 wherein said mutation co-segregates with APECED and is
- (i) an insertion;
 - (ii) a deletion;
 - (iii) a substitution; and/or
 - (iv) an inversion,
- and wherein said mutation further results in a loss of function or a gain of function of the (poly)peptide as defined in any one of claims 1 to 3.
8. The nucleic acid molecule of claim 7, wherein said insertion, which is a duplication of 4 nucleotides (CCTG) normally found at position 1086-1089, is a 4 nucleotide insertion at the nucleotide position 1085 or 1090, an insertion of an adenosine at position 1284, or an insertion of a cytosine at position 1365 of the nucleotide sequence of Fig. 2A.
9. The nucleic acid molecule of claim 7, wherein said deletion is a 13 nucleotide deletion of nucleotides 1085 -1097, a deletion of the thymidine at position 1051 or a deletion of the cytosine at position 1309 or 1313 of the nucleotide sequence of Fig. 2A.

10. The nucleic acid molecule of claim 7, wherein said substitution is a cytosine to thymidine exchange at nucleotide position 889, a guanosine to thymidine exchange at nucleotide position 358, an adenosine to guanosine exchange at nucleotide position 374, a guanosine to adenosine exchange at nucleotide position 1052, or a cytosine to adenosine exchange at nucleotide position 1094 of the nucleotide sequence of Fig. 2A.
11. The nucleic acid molecule of any one of claims 7 to 10, wherein said loss of function is a loss of macromolecule binding properties.
12. The nucleic acid molecule of any one of claims 7 to 10, wherein said gain of function is involved in molecular interaction.
13. A fragment of the nucleic acid molecule of any one of claims 1 to 12 comprising at least 14 nucleotides.
14. A nucleic acid molecule which is complementary to the nucleic acid molecule of any one of claims 1 to 13.
15. The nucleic acid molecule of any one of claims 1 to 14 which is DNA or RNA.
16. A primer pair which hybridizes under stringent conditions to the nucleic acid molecule of any one of claims 1 to 15.
17. A vector comprising the nucleic acid molecule of any one of claim 1 to 16.
18. A host transformed with the vector of claim 17.
19. The host of claim 18 which is a bacterium, a yeast cell, an insect cell, a fungal cell, a mammalian cell, a plant cell, a transgenic animal or a transgenic plant.

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20. A process of producing a (poly)peptide as defined in any one of claims 1 to 13 or 15 comprising culturing or raising the host of claim 18 or 19 and isolating said (poly)peptide from said culture or said host.
21. A (poly)peptide encoded by the nucleic acid molecule of any one of claims 1 to 13 or 15 or produced by the process of claim 20.
22. A compound derived from the (poly)peptide of claim 21 and having essential the same three dimensional structure thereof.
23. An antibody that specifically recognizes the (poly)peptide of claim 21 and/or the compound of claim 22.
24. A pharmaceutical composition comprising the nucleic acid molecule of any one of claims 1 to 15, the primer pair of claim 16, the vector of claim 17, the (poly)peptide of claim 21, the compound of claim 22 and/or the antibody of claim 23.
25. A diagnostic composition comprising the nucleic acid molecule of any one of claims 1 to 15, the primer pair of claim 16, the vector of claim 17, the (poly)peptide of claim 21, the compound of claim 22 and/or the antibody of claim 23.
26. A method for testing for a carriership for APECED or for a corresponding disease state comprising testing a sample obtained from a prospective patient or from a person suspected of carrying a predisposition for a mutation in the nucleic acid molecule of any one of claims 1 to 3.
27. A method for testing for a carriership for APECED or for a corresponding disease state comprising testing a sample obtained from a prospective patient or from a person suspected of carrying a predisposition for a mutated form of the polypeptide as defined in any one of claims 1 to 3 in an immuno-assay using the antibody of claim 23.

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28. Use of the nucleic acid molecule of any one of claims 1 to 6 or the vector of claim 17 in gene therapy.

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